

Molecular pathology: Making it simple

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UEG Week Berlin 2025

Presentation

Colorectal

Digestive Oncology

Mechanisms & Personalised Medicine

October 6, 2025

This educational presentation explains the role of molecular pathology in colorectal cancer, focusing on microsatellite instability testing to identify Lynch syndrome and predict immunotherapy eligibility.

KEY POINTS

- The speaker describes how mismatch repair protein loss, particularly mlh1, leads to microsatellite instability, which makes tumours eligible for immunotherapy as stated in the presentation.
- Immunohistochemistry staining of mismatch repair proteins provides results in a couple of hours and shows brown staining when present or blue staining when absent.
- The speaker states that approximately 30% of tumours with mlh1 hypermethylation lack Braf mutation, so hypermethylation testing should still be performed if Braf testing is negative to distinguish Lynch syndrome from sporadic cases.
- Around age 55 there is a switch where mlh1 loss before this age is mostly due to Lynch syndrome, while after 55 it is more frequently due to hypermethylation in elderly females with right-sided colon cancers.
- The speaker notes that only up to 20% of patients develop metastatic disease requiring extensive molecular testing, making universal upfront testing less cost-effective unless costs are no objection.
- Mucinous carcinomas have a 30 to 40% risk of microsatellite instability, and lymphoid aggregates visible on histology are indicative of these immunogenic tumours.

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